ARFGEF2 gene

ADP ribosylation factor guanine nucleotide exchange factor 2

Normal Function

The *ARFGEF2* gene provides instructions for making a protein that helps with the movement (trafficking) of small sac-like structures (vesicles) within the cell. The ARFGEF2 protein converts a molecule called guanine diphosphate (GDP) to another molecule called guanine triphosphate (GTP). This reaction activates an ADP-ribosylation factor, a molecule that is involved in vesicle trafficking. Vesicles transport many types of molecules from the interior of the cell to its surface, where they may attach and interact with other substances, or be secreted by the cell.

Health Conditions Related to Genetic Changes

periventricular heterotopia

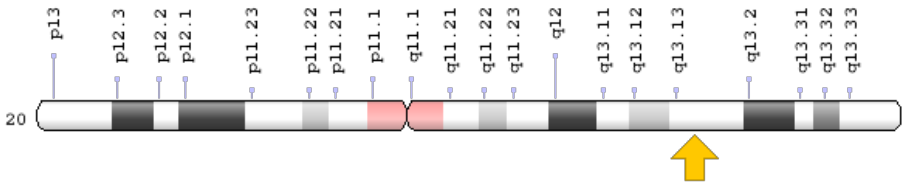
A few mutations in the *ARFGEF2* gene have been identified in individuals with periventricular heterotopia. These mutations may interfere with vesicle trafficking, which is important in controlling cell migration during the development of the brain. Nerve cells (neurons) that do not migrate properly during development form clumps around fluid-filled cavities (ventricles) near the center of the brain, resulting in the signs and symptoms of periventricular heterotopia.

Mutations in the *ARFGEF2* gene may also result in weakening of the attachments (adhesion) between cells that form the lining of the ventricles, by impairing the trafficking of the molecules needed for this adhesion. A weakened ventricular lining could allow some neurons to form clumps around the ventricles while others migrate normally to the exterior of the brain, as seen in periventricular heterotopia.

Chromosomal Location

Cytogenetic Location: 20q13.13, which is the long (q) arm of chromosome 20 at position 13.13

Molecular Location: base pairs 48,921,721 to 49,036,693 on chromosome 20 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ADP-ribosylation factor guanine nucleotide-exchange factor 2
- ADP-ribosylation factor guanine nucleotide-exchange factor 2 (brefeldin A-inhibited)
- BIG2
- BIG2_HUMAN
- brefeldin A-inhibited guanine nucleotide-exchange protein 2
- dJ1164I10.1
- FLJ23723

Additional Information & Resources

Educational Resources

- The Cell: A Molecular Approach (second edition, 2000): The Mechanism of Vesicular Transport
  https://www.ncbi.nlm.nih.gov/books/NBK9886/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28ARFGEF2%5BTIAB%5D%29+OR+%28BIG2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- ADP-RIBOSYLATION FACTOR GUANINE NUCLEOTIDE EXCHANGE FACTOR 2
  http://omim.org/entry/605371

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ARFGEF2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ARFGEF2%5Bgene%5D
- HGNC Gene Family: A-kinase anchoring proteins
  http://www.genenames.org/cgi-bin/genefamilies/set/396
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=15853
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/10564
- UniProt
  http://www.uniprot.org/uniprot/Q9Y6D5

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https://ghr.nlm.nih.gov/gene/ARFGEF2

Reviewed: November 2007
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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